# intestinal pseudo-obstruction

Intestinal pseudo-obstruction is a condition characterized by impairment of the muscle contractions that move food through the digestive tract. The condition may arise from abnormalities of the gastrointestinal muscles themselves (myogenic) or from problems with the nerves that control the muscle contractions (neurogenic).

When intestinal pseudo-obstruction occurs by itself, it is called primary or idiopathic intestinal pseudo-obstruction. The disorder can also develop as a complication of another medical condition; in these cases, it is called secondary intestinal pseudo-obstruction.

Intestinal pseudo-obstruction leads to a buildup of partially digested food in the intestines. This buildup can cause abdominal swelling (distention) and pain, nausea, vomiting, and constipation or diarrhea. Affected individuals experience loss of appetite and impaired ability to absorb nutrients, which may lead to malnutrition. These symptoms resemble those of an intestinal blockage (obstruction), but in intestinal pseudo-obstruction no blockage is found.

Some people with intestinal pseudo-obstruction have bladder dysfunction such as an inability to pass urine. Other features of this condition may include decreased muscle tone (hypotonia) or stiffness (spasticity), weakness in the muscles that control eye movement (ophthalmoplegia), intellectual disability, seizures, unusual facial features, or recurrent infections.

Intestinal pseudo-obstruction can occur at any time of life. Its symptoms may range from mild to severe. Some affected individuals may require nutritional support. Depending on the severity of the condition, such support may include nutritional supplements, a feeding tube, or intravenous feedings (parenteral nutrition).

## Frequency

Primary intestinal pseudo-obstruction is a rare disorder. Its prevalence is unknown. The prevalence of secondary intestinal pseudo-obstruction is also unknown, but it is believed to be more common than the primary form.

## Genetic Changes

In some individuals with primary intestinal pseudo-obstruction, the condition is caused by mutations in the *FLNA* gene. This gene provides instructions for producing the protein filamin A, which helps build the network of protein filaments (cytoskeleton) that gives structure to cells and allows them to change shape and move. Filamin A

attaches (binds) to another protein called actin and helps it form the branching network of filaments that make up the cytoskeleton.

Some individuals with primary intestinal pseudo-obstruction have *FLNA* gene mutations that result in an abnormally short filamin A protein. Others have duplications or deletions of genetic material in the *FLNA* gene. Researchers believe that these genetic changes may impair the function of the filamin A protein, causing abnormalities in the cytoskeleton of nerve cells (neurons) in the gastrointestinal tract. These abnormalities interfere with the nerves' ability to produce the coordinated waves of muscle contractions (peristalsis) that move food through the digestive tract.

Deletions or duplications of genetic material that affect the *FLNA* gene can also include adjacent genes on the X chromosome. Changes in adjacent genes may account for some of the other signs and symptoms that can occur with intestinal pseudo-obstruction.

Secondary intestinal pseudo-obstruction may result from other disorders that damage muscles or nerves, such as Parkinson disease, diabetes, or muscular dystrophy. Additionally, the condition is a feature of an inherited disease called mitochondrial neurogastrointestinal encephalopathy disease (MNGIE disease) that affects the energy-producing centers of cells (mitochondria). Infections, surgery, or certain drugs can also cause secondary intestinal pseudo-obstruction.

In some affected individuals, the cause of intestinal pseudo-obstruction is unknown. Studies suggest that in some cases the condition may result from mutations in other genes that have not been identified.

## Inheritance Pattern

Intestinal pseudo-obstruction is often not inherited. When it does run in families, it can have different inheritance patterns.

Intestinal pseudo-obstruction caused by *FLNA* gene mutations is inherited in an X-linked recessive pattern. The *FLNA* gene is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Intestinal pseudo-obstruction can also be inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In other families it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with

an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

In some cases, the inheritance pattern is unknown.

## Other Names for This Condition

- chronic idiopathic intestinal pseudo-obstruction
- CIIP
- congenital short bowel syndrome
- enteric neuropathy
- IPO
- paralytic ileus
- pseudointestinal obstruction syndrome
- pseudoobstructive syndrome

## Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Intestinal pseudoobstruction neuronal chronic idiopathic X-linked
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1848221/
- Genetic Testing Registry: Natal teeth, intestinal pseudoobstruction and patent ductus
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855732/
- Genetic Testing Registry: Visceral myopathy familial with external ophthalmoplegia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1848586/
- Genetic Testing Registry: Visceral neuropathy familial
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855733/
- Genetic Testing Registry: Visceral neuropathy, familial, autosomal dominant
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864996/

Other Diagnosis and Management Resources

- Children's Hospital of Pittsburgh
  http://www.chp.edu/our-services/transplant/intestine/education/intestine-disease-states/chronic-intestinal-pseudo-obstruction

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

## Additional Information & Resources

MedlinePlus

- Encyclopedia: Primary Intestinal Pseudoobstruction
  https://medlineplus.gov/ency/article/000253.htm
- Health Topic: Colonic Diseases
  https://medlineplus.gov/colonicdiseases.html

Genetic and Rare Diseases Information Center

- Intestinal pseudo-obstruction
  https://rarediseases.info.nih.gov/diseases/6789/intestinal-pseudo-obstruction

Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: Intestinal Pseudoobstruction
  https://www.niddk.nih.gov/health-information/digestive-diseases/intestinal-pseudo-obstruction

Educational Resources

- Children's Hospital of Pittsburgh
  http://www.chp.edu/our-services/transplant/intestine/education/intestine-disease-states/chronic-intestinal-pseudo-obstruction
- Disease InfoSearch: Intestinal pseudo-obstruction
  http://www.diseaseinfosearch.org/Intestinal+pseudo-obstruction/3848
- MalaCards: intestinal pseudo-obstruction
  http://www.malacards.org/card/intestinal_pseudo_obstruction

Patient Support and Advocacy Resources

- Association of Gastrointestinal Motility Disorders
  http://www.agmd-gimotility.org/
- International Foundation for Functional Gastrointestinal Disorders
  http://aboutkidsgi.org/intestinal-pseudo-obstruction.html
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/chronic-intestinal-pseudo-obstruction/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22intestinal+pseudo-obstruction+syndrome%22+OR+%22Intestinal+Pseudo-Obstruction%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Intestinal+Pseudo-Obstruction%5BMAJR%5D%29+AND+%28intestinal+pseudo-obstruction+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- INTESTINAL PSEUDOOBSTRUCTION WITH PATENT DUCTUS ARTERIOSUS AND NATAL TEETH
  http://omim.org/entry/243185
- INTESTINAL PSEUDOOBSTRUCTION, NEURONAL, CHRONIC IDIOPATHIC, X-LINKED
  http://omim.org/entry/300048
- VISCERAL MYOPATHY, FAMILIAL, WITH EXTERNAL OPHTHALMOPLEGIA
  http://omim.org/entry/277320
- VISCERAL NEUROPATHY, FAMILIAL, AUTOSOMAL DOMINANT
  http://omim.org/entry/609629
- VISCERAL NEUROPATHY, FAMILIAL, AUTOSOMAL RECESSIVE
  http://omim.org/entry/243180

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/intestinal-pseudo-obstruction

Reviewed: October 2010
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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